



TYMP gene

thymidine phosphorylase

Normal Function

The *TYMP* gene (previously known as *ECGF1*) provides instructions for making an enzyme called thymidine phosphorylase. Thymidine is a molecule known as a nucleoside, which (after a chemical modification) is used as a building block of DNA. Thymidine phosphorylase converts thymidine into two smaller molecules, 2-deoxyribose 1-phosphate and thymine. This chemical reaction is an important step in the breakdown of thymidine, which helps regulate the level of nucleosides in cells.

Thymidine phosphorylase plays an important role in maintaining the appropriate amount of thymidine in cell structures called mitochondria. Mitochondria convert the energy from food into a form that cells can use. Although most DNA is packaged in chromosomes within the nucleus, mitochondria also have a small amount of their own DNA (called mitochondrial DNA or mtDNA). Mitochondria use nucleosides, including thymidine, to build new molecules of mtDNA as needed.

Health Conditions Related to Genetic Changes

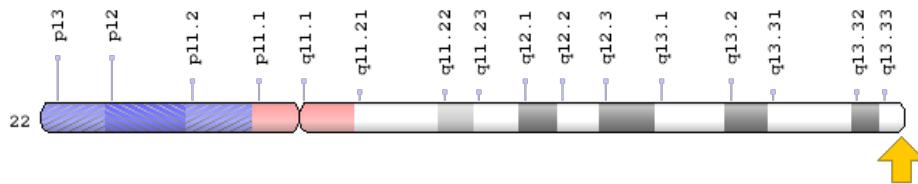
mitochondrial neurogastrointestinal encephalopathy disease

About 50 mutations in the *TYMP* gene have been identified in people with mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease. *TYMP* mutations greatly reduce or eliminate the activity of thymidine phosphorylase. A shortage of this enzyme allows thymidine to build up to very high levels in the body. An excess of thymidine appears to be damaging to mtDNA, disrupting its usual maintenance and repair. As a result, mutations can accumulate in mtDNA, causing it to become unstable. Mitochondria may also have less mtDNA than usual (mtDNA depletion). These genetic changes impair the normal function of mitochondria. Although mtDNA abnormalities underlie the digestive and neurological problems characteristic of MNGIE disease, it is unclear how defective mitochondria cause the specific features of the disorder.

Chromosomal Location

Cytogenetic Location: 22q13.33, which is the long (q) arm of chromosome 22 at position 13.33

Molecular Location: base pairs 50,525,752 to 50,530,085 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ECGF1
- endothelial cell growth factor 1 (platelet-derived)
- gliostatin
- hPD-ECGF
- MNGIE
- PD-ECGF
- PDECGF
- TdRPase
- TP
- TYPH_HUMAN

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): The Genetic System of Mitochondria
<https://www.ncbi.nlm.nih.gov/books/NBK9896/#A1629>

GeneReviews

- Mitochondrial Neurogastrointestinal Encephalopathy Disease
<https://www.ncbi.nlm.nih.gov/books/NBK1179>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TYMP+NOT+tympanometry+NOT+otitis%5BTIAB%5D%29+OR+%28thymidine+phosphorylase%5BTIAB%5D%29+OR+%28ECGF1%5BTIAB%5D%29+OR+%28MNGIE%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- THYMIDINE PHOSPHORYLASE
<http://omim.org/entry/131222>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/TYMPID40397ch22q13.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TYMP%5Bgene%5D>
- HGNC Gene Family: Minor histocompatibility antigens
<http://www.genenames.org/cgi-bin/genefamilies/set/870>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3148
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1890>
- UniProt
<http://www.uniprot.org/uniprot/P19971>

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